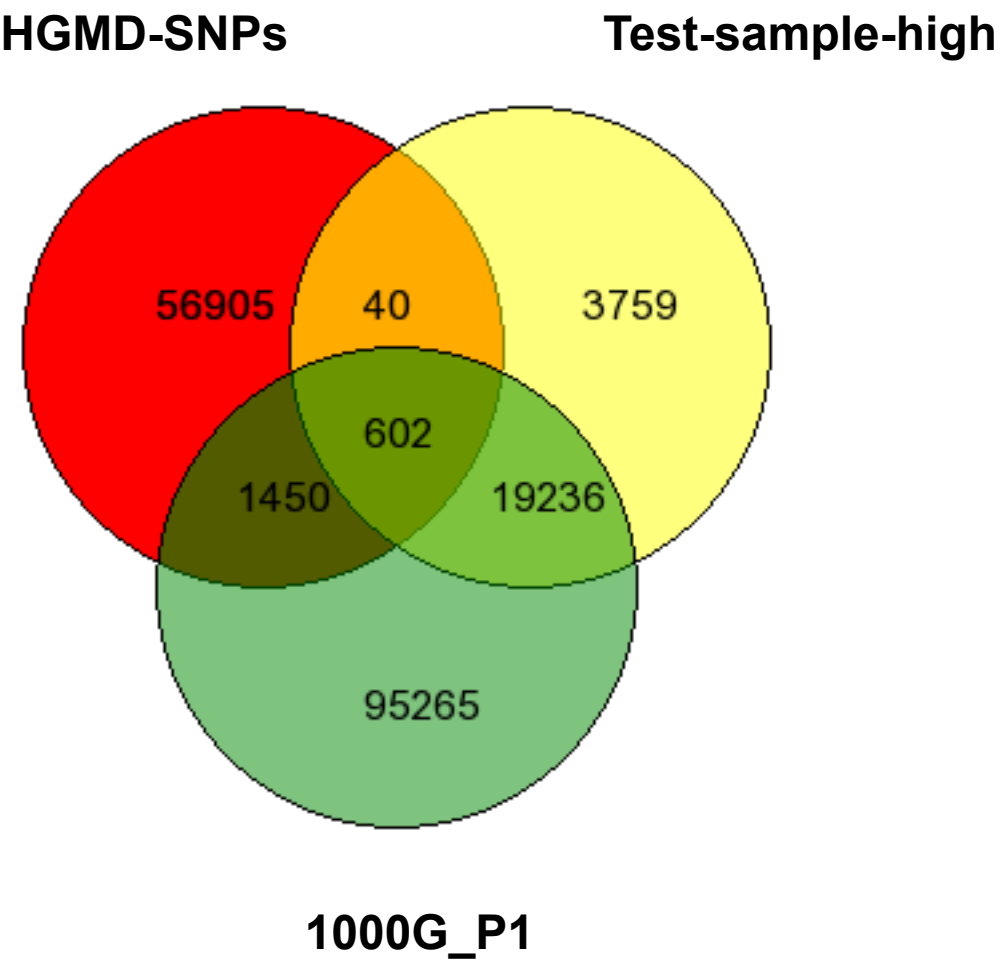
Mapping based on GENCODE v3c

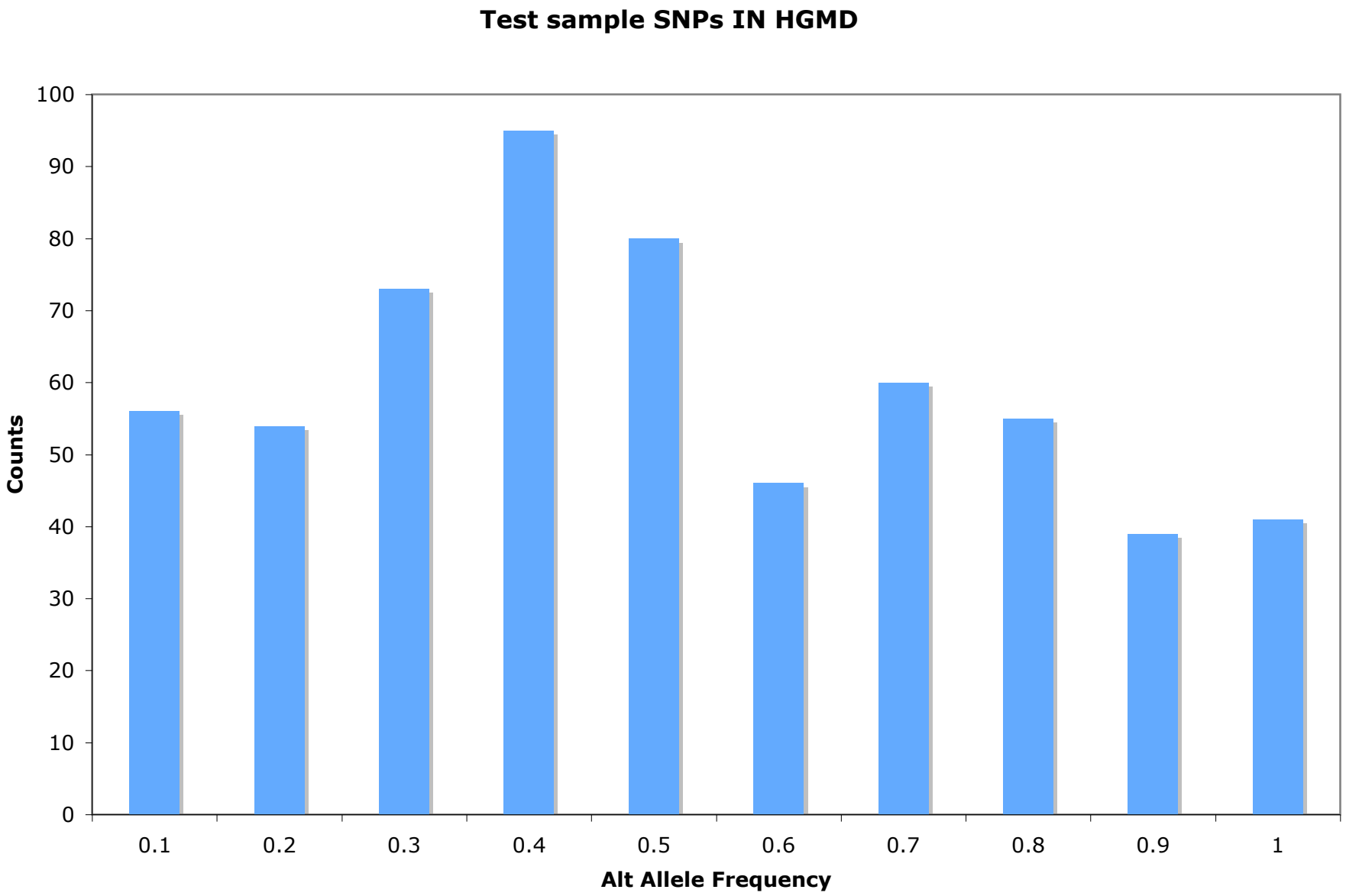
Dataset	High conf	Refseq (annovar)	Low conf
Total	3,301,521		438,180
Intronic	1,092,522 (509,328)	1,241,815	124,390 (58,684)
Nsyn	12,216 (6425)	10,741	1,566 (1031)
Syn	12,129 (6350)		914(587)
Premature	148 (62)	98	31 (23)
Stop removed	37 (0)		2 (0)
Splice	123 (41)		37 (21)

SNPmapper developed by Lukas H

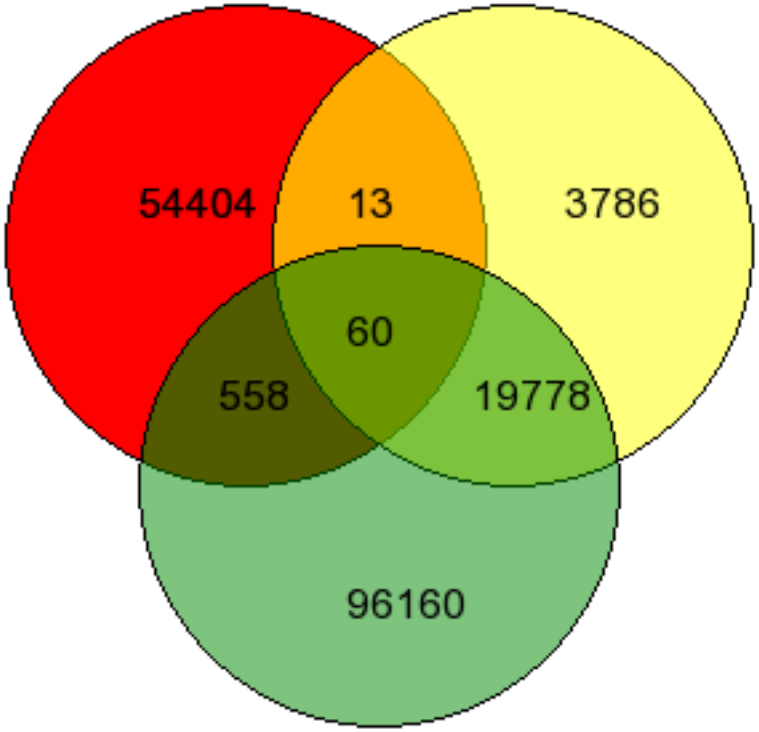
The numbers in the parentheses indicates number of cases where the SNP is present in all isoforms of a transcript

Coding SNPs intersected with HGMD mutations

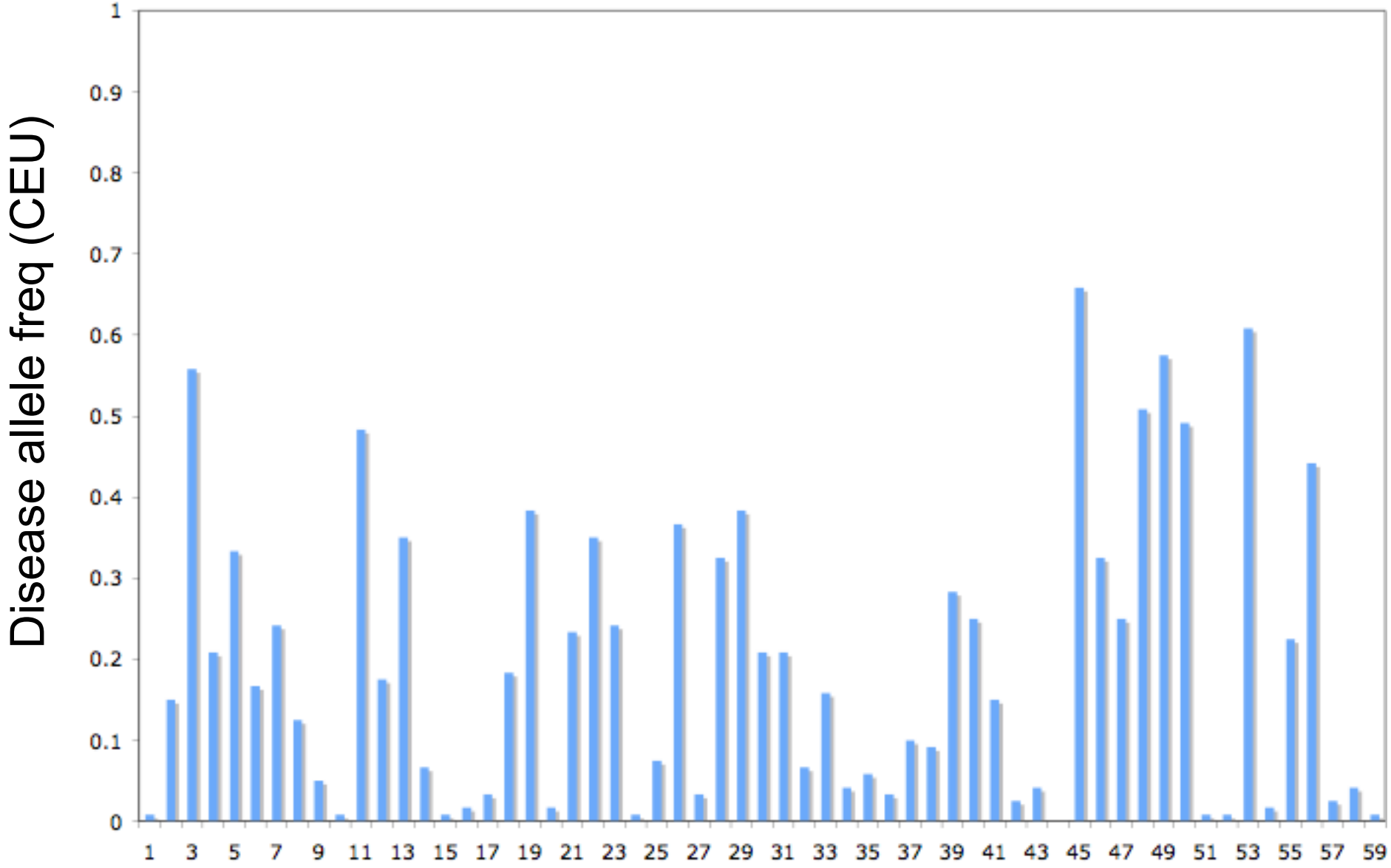


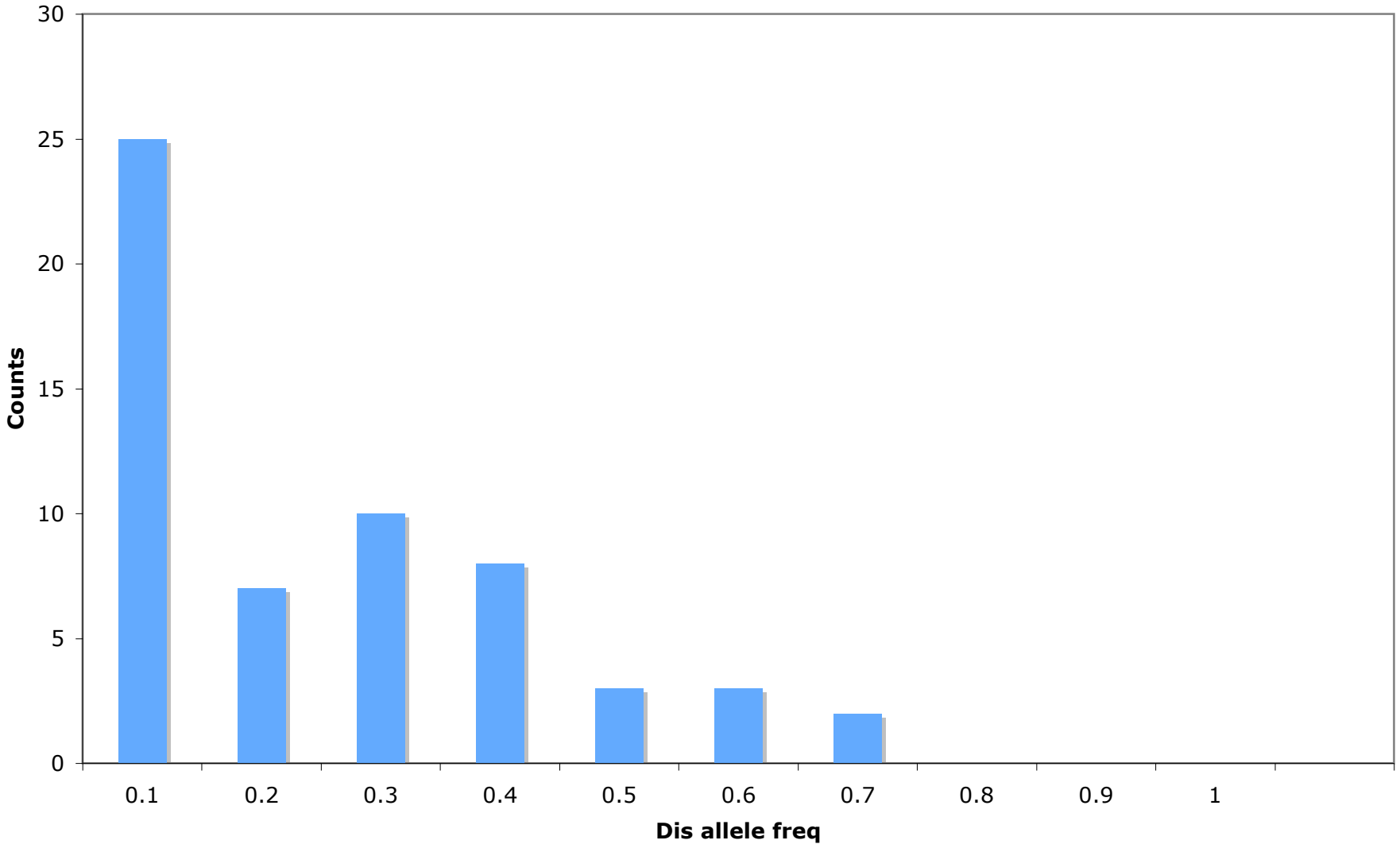

Test sample SNPs IN HGMD
Counts
100
90
80
70
60
50
40
30
20
10
0
0.1
0.2
0.3
0.4
0.5
0.6
0.7
0.8
0.9
1
Alt Allele Frequency

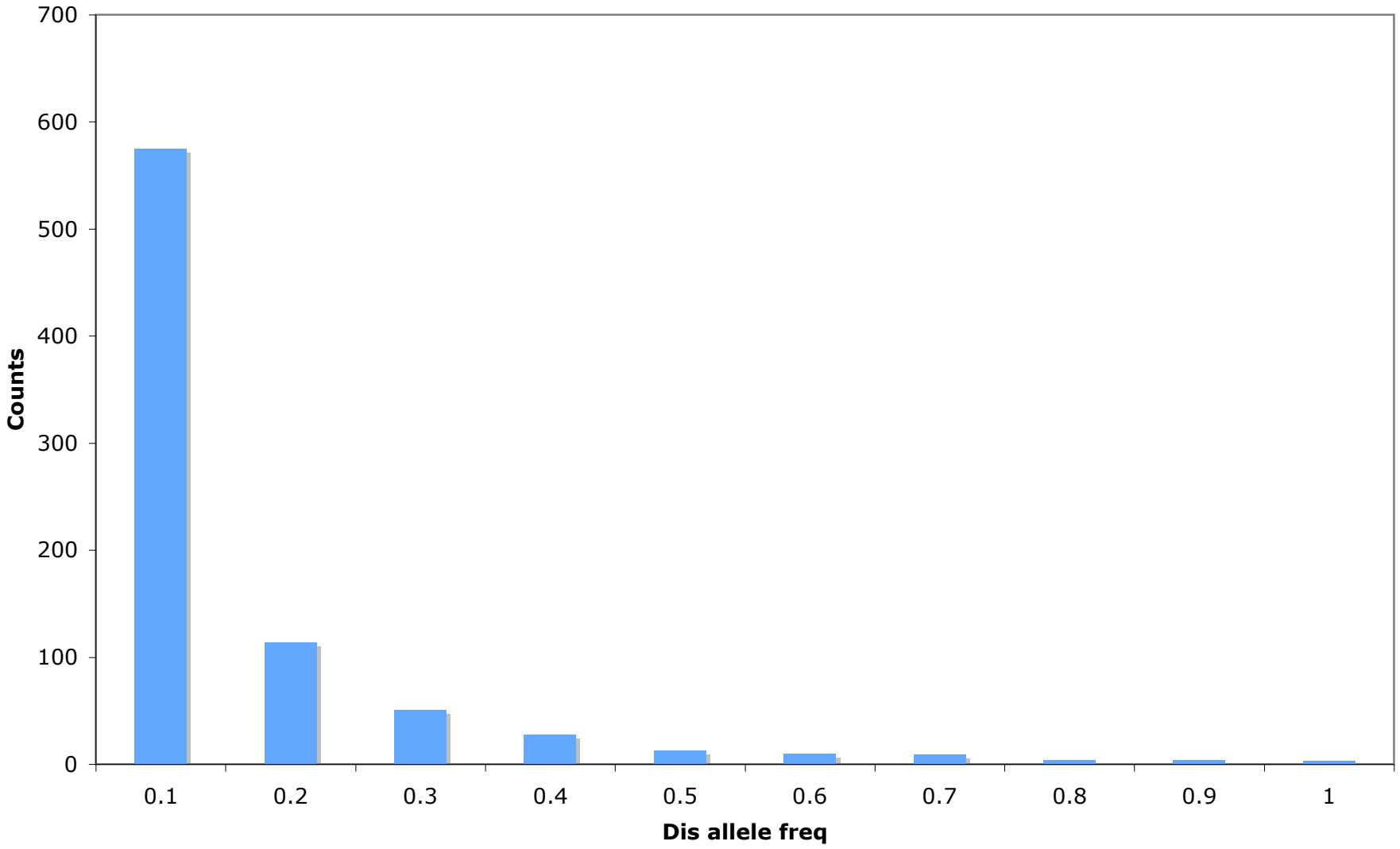
HGMD mutations in P1 and test sample



HGMD disease mutations in P1 and test sample




HGMD SNPs in test sample and P1
30
25
20
15
10
5
0
Counts
0.1
0.2
0.3
0.4
0.5
0.6
0.7
0.8
0.9
1
Dis allele freq


P1 SNPs in HGMD
700
600
500
400
300
200
100
0
Counts
0.1
0.2
0.3
0.4
0.5
0.6
0.7
0.8
0.9
1
Dis allele freq